



Osteogenesis Imperfecta
Federation Europe

Newsletter

Issue 28, March 2016

In This Issue:

Editorial	P. 1-2
What is the OIFE doing?	P. 2-3
Adult services for OI – What happens in Omaha?	P. 4-7
News in brief	P. 8-9
Calendar + Contact	P. 9-10

Editorial

Dear readers,

It seems like most of you welcomed in a very positive and warm way the news about the upcoming OIFE YOUTH Meeting in London. The first 40 places offered by BBS (Brittle Bone Society, UK) for free went "sold out" in the blink of an eye.

I was sure this would happen. The VOICE meeting (BBS youth event) is since a while now a reference point for many of the OI European youth groups.

Furthermore this weekend offers the chance to visit one of the most popular destinations of the world!

I have a personal and deep relationship with London. I was 14 years old the first time I went there. I remember my mother panicking at the airport asking me not to move from my seat until the aircraft had completely stopped. I remember me getting to the college where I spent those 2 weeks and finding in my suitcases a couple of hoodies less (thing that gave me the chance to go for more shopping than planned) and some different size and kind of plaster cast that I am sure I had not packed myself.

I remember the feeling of being free that I had those 2 weeks, meeting new people and sharing with them awesome experiences and adventures...

I am sure everyone of you will feel similar feelings when you'll be leaving for London next month. I am also sure this will be a great, huge youth event.

During the Saturday workshops and talks program participants will also have the chance to discuss some very relevant and somehow complicated topics and issues related to OI. I am sure that this mix of fun and education can be very important and might result in being a good concept to repeat in the future youth meetings.

As Mrs Catherine Potterton, one of the founders of VOICE meeting, stated in her Declaration of Independence at the OIFE seminar in Lisbon, the self empowerment of

every one of us OIERS goes through several obstacles and paths. But the one about getting to know ourselves, our situations and learning how to deal with it in our daily life as well as when making plans for the future is really important.

So please all you participants of the upcoming OIFE Youth meeting, try to exploit as much as possible this chance for meeting, discussing, getting to know other people, new topics, new issues related to OI even when those seems to be difficult to understand or just boring to follow.

And yes, well of course and first of all do not forget to have fun!

Anna Rossi
OIFE Youth Coordinator
youth-coordinator@oife.org



What is the OIFE doing?

Here are some headlines about what the OIFE has been doing the last 3 months:

Networking and collaboration

The OIFE has attended the following meetings/conferences the last 3 months:

EC-meeting (Skype) – January 19th 2016
EURORDIS Webinar on ERNs – February 10th 2016
Skype-meeting with Care4BB – March 13th 2016

Research and pharma industry

The OIFE has received requests about collaboration with research projects and pharmaceutical companies. Latest about a planned phase 3 interventional study in adult (ages 18 or older) type I, III or IV OI patients. The study drug is a monoclonal antibody. Do you know any investigators who might be interested in collaboration regarding this? Send an e-mail to office@oife.org.

European Reference Networks (ERNs)

European Reference Networks are networks of centres of expertise, healthcare providers and laboratories that are organised across borders.

A group of European clinicians are in the process of building up a European Reference Network (ERN) on rare bone diseases with two subgroups - Skeletal Dysplasia and Metabolic Bone Diseases. The main goal of the network is to significantly increase quality of care, improving early diagnosis capabilities, to improve care in disadvantaged areas at European level and last but not least to support research. There are at the moment no initiatives regarding the group "Rare connective tissue and musculoskeletal diseases" that would also be relevant to OI.

Did you know there is a "Matchmaker" online? If you know about researchers or centres of expertise who want to be part of the process – tell them to check out <http://www.rd-action.eu/european-reference-networks-erns>

OIFE webpages were hacked!

The page www.oife.org is currently undergoing a redesigning because of serious technical issues with the old system. We will restore the website's content bit by bit. In the meantime you can check out news from the OIFE on our Facebook-pages (we want as many followers as possible): <https://www.facebook.com/OIFEPAGE>

OIFE AGM

We remind you to save the date for the next OIFE AGM taking place in Lisbon from October 9th–10th (meeting on Sunday AND Monday) after the 1st Hispanoluso Congress about OI, which takes place from October 7th - 8th.



SAVE the DATE **OI in 2016**

Latest Developments in Osteogenesis Imperfecta

7^{to}8 **Lisbon**
October **Portugal**

1st Hispanoluso Congress on
Osteogenesis Imperfecta

UN breakable Alliance  FUNDACIÓN 
associação portuguesa de osteogénese imperfeita Osteogénese imperfeita

more information: info.fundacion@ahuce.org
comunicacao@apoi.pt

Adult services for OI – what happens in Omaha?

Ingunn Westerheim – OIFE president

Adult services for OI has been an important topic for me resulting in the seminar “Soft Tissues & Soft Issues – OI in adults is more than fractures” in 2015. Before this, I took part in the big adult population study in Norway with almost 100 participants. The study showed that the challenges adults with OI face are different from the ones they struggle with as children. NFOI has been working for better services and follow-up routines for adults with OI after the study. If we know what and when to check –we will live even healthier and more productive lives than we already do.

The TRS Centre (National Resource Centre for Rare Disorders, Norway) has published a guide for GPs (follow up routines) in 2016. So far it is only available in Norwegian, but we are trying to get it translated into English. And Norway is not the only country working with adults with OI. The Netherlands has had an adult clinic with OI for a long time, the UK is developing adult services and the US just launched their adult clinic in Omaha, Nebraska.

And there seems to be a lot going on in Omaha these days. We have asked Dr. Desai and Dr. Rush to tell us a little bit about their work to establish a clinic for adults with OI and why this all started:

Dr. Desai: *Well, the easiest answer is "because there is a need". Children in the US who age out of Pediatric OI clinic will be forced to find care on their own once they are an adult, and currently there are very few places,if any, that offer comprehensive multidisciplinary adult OI care services. Since adults with OI need care as well, it only made sense that this clinic could serve both populations.*

Dr. Desai – you have said “There seemed to be more focus on diagnosing and researching OI then actually clinically managing OI.” What do you mean?

Dr. Desai: *There are different aspects of the medical community and they each serve a purpose. Some are in research labs, studying animal models of OI to understand the genetics, and how OI develops at a microscopic level. Some are doing clinical trials of investigative medications. Others are clinically managing patients with OI. This last group is the group that is lacking in my opinion. I'm a family physician - I manage many chronic conditions like hypertension, diabetes, asthma, hypothyroidism, and hyperlipidemia just to name a few. I know these conditions affect each patient individually and different therapies in treating them and when to use them, potential side effects, appropriate diagnostic testing and timing of when to do these things. You just don't see that with OI. That's clinical management.*



Can you tell us a little bit of your background and involvement in OI work?

Dr. Rush: *I am a clinical geneticist who was previously trained in Internal Medicine and Paediatrics. When I started my fellowship in genetics, it was suggested to me by a colleague that since I was looking for a research project, perhaps I could find one that would make use of the large numbers of OI patients seen in Omaha. I started a small research project, and then just started attending clinic because I enjoyed working with the patients and families. Three years ago, I took over as medical director of our OI clinic in Omaha. I have been privileged to work with a wonderful group of families who have been incredibly supportive of our clinic.*

Dr. Desai: *I am a family physician and have a 6 year old son with OI. He was diagnosed with OI when my wife was pregnant with him. Initially, the first doctor said he wouldn't live. He did. Then doctors said he wouldn't eat. He did. Then doctors said he wouldn't breathe. He did. Then doctors said he wouldn't walk or run. He did those things too. As a doctor, I trusted many of my medical colleagues, because I thought they knew about OI and were telling me the truth. I found the opposite was true, and the only way I could trust any doctor ever again was to learn myself as much as I could about OI. This eventually led me to forming a strong friendship and partnership with Dr. Eric Rush in Omaha. Their approach to OI care, compassion, and experience (of treating almost 200 kids with OI since 2002) was the reason I joined with them in forming an organization with their Children's Foundation.*

Your name shows up in many different settings these days, Dr. Rush - do you have plans on becoming the next "OI guru"?

Dr. Rush: *I have every intention of working with people who have OI as a major focus of my career as long as I stay in good health. However, I would much rather do so as part of a collaborative group of engaged patients, clinicians, investigators, and advocacy groups.*

How did the first OI clinic for adults go?

Dr. Rush: *It went well. I hope that our patients felt that the clinic was of benefit to them. As with all things, we have identified opportunities for further innovation and*

better coordination. I have an excellent group of case managers who have really taken to their roles in this clinic and have proved indispensable. Our paediatric OI clinic has been constructed such that most of the moving parts are present on the day of clinic and we have opportunities to meet face-to-face on that day. It's a good way to do things, but is fairly resource intensive. Realities of adult medicine would appear to preclude an identical model, so we will have patients see most providers individually. As long as we are providing a high level of coordination and support for this care, I think this is still of benefit.

What are your further plans regarding the clinic?

Dr. Rush: *One of the most exciting aspects of this clinic is that it will alter organically as we discover more about the natural history of OI. I'm really excited about this clinic not only providing the best quality care, but also being a testing laboratory for different care models. The overarching principle of this clinic is one of collaborative care. Collaborative care is very different from the focus on "customer service" that is plaguing an increasingly corporatized health care system in the United States.*

Why should there be separate clinics for adults? Why can't they just continue to be seen by the paediatrician? Or followed by their local GP?

Dr. Desai: *This is just the way pediatric and adult care is provided in the United States. Pediatricians treat children, usually until around 18 or 21, then an adult physician takes over for adulthood. Adults should still have a local GP, but with so many different aspects of OI care, it helps to have a more specialized center familiar with OI to help guide both the local GP and adult with OI who may not know everything about OI care.*

Dr. Rush: *Everyone with OI should have a general practitioner/primary care provider that they have good rapport with and who continues to provide the majority of care. Most of the primary care that patients with OI require are part of being human, and we should resist the temptation to define a person entirely by their rare disease. I find that patients with rare diseases often stay with a paediatric provider for longer in their lifespan, which is a testament to the quality of care that is provided. We have a saying in paediatrics that "children are not just small adults". This is a true statement, but a corollary exists in that "adults are not just large children". An overlapping set of issues exists in our adult population, and we need to be prepared to engage patients on subjects such as chronic pain management, post-secondary education and employment, sexual health and family planning, and the effects of aging on patients with OI. This requires a different set of medical providers for adult patients than we utilize for paediatric patients, and probably a different care model.*

What kind of professionals should a clinic for adults with OI have and why?

Dr. Rush: *This is a good question and one that I don't think we have completely*

answered yet. I think there has to be a focal point with any clinic of this type, but what that person looks like will be different based on the location. They certainly do not need to have identical training to mine. I would say that training in adult medicine with interest and experience in OI is a must. I think some patients will require the services of cardiology, nephrology, and pulmonology. Patients with problems of chronic pain may benefit from management by a pain medicine specialist. Orthopaedic surgeons may be of assistance to patients to address specific joint-related concerns. I also suggest routine surveillance by an ophthalmologist as patients with OI appear to have an increased risk of eye health concerns, and diagnosis may be more difficult in patients with OI due to some changes to the structure of the eye (e.g. thin corneas contribute to incorrect intraocular pressure readings). This list of providers will undoubtedly change over time.

Dr. Desai: *This is something we are starting to develop in Omaha - it is going to vary, and I can not make any specific recommendations, but what I can do is give you examples of who we will be involving in Omaha: Cardiology, Pulmonology, Orthopedics, Spine, Physical Therapy, Ophthalmology, Audiology, Nephrology, Genetics (Dr. Rush), Occupational Therapy, Social Worker, and then will refer to additional specialists as needed on case by case basis. These specialists are going to be valuable in caring for the OI patient as OI affects so many different organ systems and not just bones. We are just starting the clinic, so these are some of specialists we are targeting. Different centers should feel free to have specialists as they see fit, depending on the needs of their community and resources available. This is by no means the only way to have an OI clinic, but we think it helps with some basic OI care model guidelines others can follow. And of course, we are always happy to consult and give our experiences as well.*

Dr. Desai -You have established a group on Facebook called “Dr. D’s OI Clinical Resource Group”. Who is it for? And what is the purpose behind the group?

Dr. Desai: *I just think it's a great way to have an online community of OI providers, parents, advocates, and adults with OI to exchange ideas and learn together - very collaborative as opposed to having a "specialist" with an ego dictate everything to everyone else. It has been very successful so far I think, and I hope to grow it for a long time.*

Thank you Dr. Desai and Dr. Rush for the interview.

News in brief

"**The HOI foundation** in the Netherlands has sent a brandnew electric wheelchair to Fathima Nyas in Ninthavur, Sri Lanka. We were able to find a company who was willing to adapt the chair for free.

By the end of January the electric wheely has been picked up by my house and brought to the world harbour in Rotterdam. It will go by containership to Colombo, the capital of Sri Lanka, where it shall be taken over by our contact persons there.

So Fathima will have more freedom in her life and be able to visit school by herself!"

Hinke Panjer
HOI Foundation Netherlands
Hulp aan mensen met **O**steogenesis **I**mperfecta
Website: www.hoistichting.nl



Fathima has been waiting for an electric wheelchair for a long time.

Announcement: The BBS and OIFE youth meeting 2016 will take place in London!

There will be loads of fun activities, workshops and a disco.

Where – London, Radisson Blu Hotel, Stansted airport

When – Friday 29th April – Sunday 1st May

Who – you and all your OI friends (aged 16-30)

The 40 first free places are taken. Remaining places (with higher fees) are limited – but people who are interested can register here :

<http://www.brittlebone.org/lat.../voice-oife-youth-event-2016/>



CONGRATULATIONS!

Lucas from Spain is one of the winners of the EURORDIS (European Organisation for Rare Diseases) photo contest 2015.

The winning photo, as voted for by the public, shows Lucas who is living with OI. After recently fracturing his arm, he still did not miss the annual parade of the historic Cantabrian Wars!

CONTACT REGISTRY – Brittle Bone Disorders Consortium: The Brittle Bone Disorders Consortium (USA) is a group of physicians, researchers, and educators focused on learning more about OI and developing new and better treatments to improve the care of patients with OI. Did you know that people from other countries than the US, can also join the Rare Diseases Clinical Research Network Contact Registry for the Brittle Bone Disorders Consortium (Contact Registry)?

Go to www.rarediseasesnetwork.org/cms/BBD

Calendar

2016

April 8-9: Meeting for adults DFOI & AGM (with family meeting) – DFOI (Denmark)

April 29 – May 1st: AGM & family meeting NFOI (Norway)

May 6th : Wishbone Day (international awareness day for Osteogenesis Imperfecta)

May 26-29: Annual General Meeting with OI Conference DOIG in Duderstadt, Germany

June 4-5: 30th anniversary SVOI (Swiss OI-Association)

July 23-30: Family Camp DFOI (Denmark)

July 22-24: OIF National Conference, Walt Disney World Swan and Dolphin Resort,
Orlando, Florida www.oif.org/conference

July 28-31: Family camp at Geilo (Norwegian OI association NFOI)

September 22-25: OI-meeting adults NFOI with medical TRS-seminar

October 7-10: OIFE Annual General Meeting and 1st Hispanoluso OI Congress in
Lisbon, Portugal

October 23: Medical meeting in Leuven, Belgium (ZOI – Belgium)

2017

August 27th – 30th: 13th International Conference on Osteogenesis imperfecta in Oslo,
Norway – www.oioslo2017.org

Contact

Ingunn Westerheim (President): president@oife.org:

Ute Wallentin (Networking&Social

Projects): socialnetwork@oife.org

Stefanie Wagner (newsletter editor and secretary):

secretary@oife.org

Anna Rossi (Youth Co-ordinator): youth-coordinator@oife.org

Website: www.oife.org

Facebook: <https://www.facebook.com/OIFEPAGE>

Twitter: @OIFE_OI